

A rare case report of Weill Marchesani syndrome.

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Abstract:

Weill Marchesani syndrome is a rare genetic disorder characterized by short stature, brachydactyly, microspherophakia, lenticular myopia, ectopia lentis and lens induced glaucoma. Ocular complications can be managed by laser iridotomy, lens extraction & trabeculectomy if needed. In this report, we present the clinical manifestations of Weill Marchesani syndrome in a 13 year old female patient presenting with diminution of vision in both eyes with similar complains in her brother. On examination she had short stature, brachydactyly, bilateral microspherophakia and pupillary block glaucoma. For both eyes clear lens extraction was performed followed by right eye trabeculectomy with mitomycin C. Her best corrected visual acuity improved to 6/24 from 6/60 in both eyes and with control of intraocular pressure (right eye 8 mm Hg from 40 mm Hg, left eye 14 mm Hg from 28 mm Hg with use of topical treatment in left eye).

Key words: Ectopia lentis, Microspherophakia, Pupillary block glaucoma.

Introduction:

Weill Marchesani syndrome is a rare connective tissue disorder occurring in 1 in 100,000 individuals^{1, 2}. It was first described by Weill and Marchesani^{3,4}. Its inheritance can be either of an autosomal recessive form which has been mapped to a locus on chromosome 19p or a phenotypically similar autosomal dominant form which is caused by mutation in the fibrillin-1 gene on chromosome 15q21⁵. Ocular abnormalities include microspherophakia, ectopia lentis, high lenticular myopia, lens induced glaucoma and corneal changes^{6,7}. Systemic abnormalities include short stature, progressive joint stiffness, brachydactyly, short extremities, thick inelastic skin, mild mental retardation and cardiac abnormalities^{8,9, 10}. A high degree of myopia with a shallow anterior chamber should alert the physician to this diagnosis, especially in young person. The small, round lens in this condition also has loose zonules, and the glaucoma may be related to lens dislocation or a forward shift of the lens, causing pupillary block glaucoma¹¹, which can be precipitated or aggravated by miotic therapy. Bilateral angle closure glaucoma has also been reported after mid-dilatation with cyclopentolate in a child with the Weill Marchesani syndrome but without lens subluxation¹². In the absence of any complication, the patient can be managed with myopic correction and regular follow up. Early detection and removal of the ectopic lens can reduce the possibility of pupillary block induced glaucoma. Angle closure glaucoma may be treated with laser iridotomy or peripheral iridoplasty depending on the relative proportion of

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pupillary block¹³. In the presence of angle closure and cataract induced visual loss, lensectomy, IOL insertion and glaucoma surgery have been described as a viable approach¹⁴.

Hereby we report a case of Weill Marchesani syndrome with bilateral lenticular myopia, microspherophakia & pupillary block glaucoma which was managed by clear lens extraction in both eyes followed by right eye trabeculectomy with mitomycin C. Timely diagnosis and appropriate management improved the visual function with control of intraocular pressure and prevented further visual handicap.

Case report

A 13 year old female from Rajasthan came to our tertiary care center with chief complains of gradual, progressive, painless diminution of vision in both eyes for 7 years. Similar complaints were present in her brother for which he was operated, details of which were not available.

Visual acuity was 6/60 with -26.00 D Sph in both eyes on the Snellen's chart. Ocular adnexa were normal. On slit lamp examination, both eyes showed iridodonesis, sluggishly reacting pupil with irregular pupillary border and microspherophakia[Image 1,2]. Intraocular pressure by Perkins applanation tonometer was 40 mm Hg in right eye and 28 mm Hg in left eye. On gonioscopic examination by Goldmann four mirror gonioscope all angles were 0 (Shaffer grade) with convex iris configuration. On ultra biomicroscopy, both eyes showed spherophakia with increased lens thickness 4.83 mm (right eye) and 4.79 mm (left eye). Central corneal thickness on ultrabiomicroscopy was 0.60 mm in right eye and 0.58 mm in left eye. Axial length on a scan was 22.88 mm in right eye and 22.98 mm in left eye. On fundus examination by direct and indirect ophthalmoscope with 20 D lens, both eyes showed 0.7-0.8 cup to disc ratio, nasalization of vessels, healthy neuroretinal rim and a normal foveolar reflex.

Physical examination showed short stature, brachycephaly, short stubby fingers and toes[Image 3-6]. Other systemic examination was normal. Blood investigations like complete blood count, random blood sugar, renal function tests and liver function tests were within normal limits.

Image 1 Slit lamp photo of right eye showing Microspherophakia with pupillary block

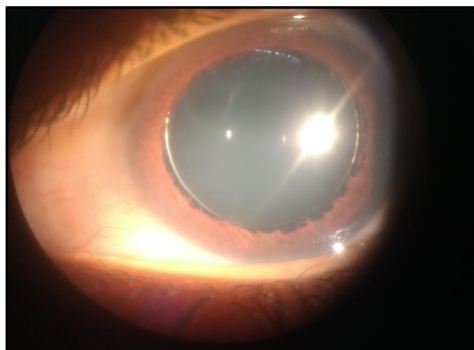
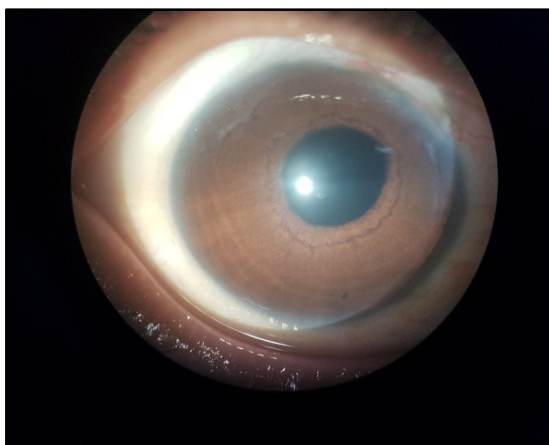


Image 2 Slit lamp photo of left eye showing Microspherophakia with pupillary block



Image 3 Short stature**Image 4 Brachydactyly****Image 5 X-ray showing Brachydactyly****Image 6 X-ray showing brachycephaly****Image 7 Slit lamp photo of Right eye post cataract surgery****Image 8 Slit lamp photo of Left eye post cataract surgery**

Diagnosis was made as Weill Marchesani syndrome with bilateral lenticular myopia, microspherophakia & pupillary block glaucoma.

In both eyes, clear lens extraction followed by right eye trabeculectomy with mitomycin C was done [Image 7,8].

On post operative day 25, visual acuity for distance in right eye was 6/24 with

+9.0DSph/+2.0 DCyl at 90° and in left eye was 6/24 with +9.0DSph on Snellen's chart. Near visual acuity in both eyes on Roman test type was N/10 with addition of +2.50D Sph. Intraocular pressure by Perkins applanation tonometer in right eye was 8 mm Hg. In left eye, the pressure was 14 mm Hg with use of brimonidine 0.2% and timolol 0.5% eye drops twice a day.

Considering similar history in brother, we educated the parents for genetic counselling.

Discussion:

Weill Marchesani syndrome is a very rare disease and hence, it often remains undiagnosed until the patient presents with complications. In his original publication, Marchesani hypothesized that an overdevelopment or hyperplasia of the ciliary body might be the reason for the spherophakia⁴. However, an ultrasonographic biomicroscopy study of three patients with this syndrome having normal axial lengths demonstrated that the ciliary body actually appeared smaller than normal¹⁵. The investigators hypothesized that the small ciliary body represents the underlying reason for elongated zonules and that it may be exerting less force on the lens, giving rise to the spherical shape of the lens. An ultrastructural study of the lens from a patient with the Weill Marchesani syndrome revealed degeneration and necrosis of the epithelial cells and destruction of cortical fibre, which was thought to result, in part, from the trauma and irritation of a highly mobile lens in close contact with the iris¹⁶. Microspherophakia may be progressive and accounts for the marked myopia observed in these patients⁸. In one study done by Jensen, Cross and Paton, myopia of -5.00 to -20.50 D was found in all the eyes measured. In our case myopia of -26 D was present in both eyes. Refractive correction required after lens extraction in these same patients was similar to that of normal aphakic eyes, confirming the lenticular origin of the myopia¹¹. The abnormal lens shape and lens dislocation into the anterior chamber can also cause pupillary block and chronic angle closure glaucoma, which has been noted to occur in up to 76% of eyes¹¹. In our case also, an abnormal lens shape had caused pupillary block glaucoma. Ectopia lens is most often central⁸. Verloes et al described a Weill Marchesani like syndrome in 3 generations of one family showing dominant inheritance. They proposed a new name GEMSS syndrome (glaucoma, ectopia, microspherophakia, stiff joints, short stature) for dominantly inherited Weill Marchesani like syndrome to distinguish it from the classical recessive Weill Marchesani syndrome¹⁷. Recently additional features of Weill Marchesani syndrome have been reported in the literature. Razeghinejad et al. analyzed the corneal thickness in 6 cases and found that the average corneal thickness was 631 ± 25.9 micron in syndromic patients, while it was 535.8 ± 25.9 micron in normal subjects⁷. By confocal examination; they revealed that this increase in corneal thickness was associated with the activation of keratocytes in the anterior stroma¹⁸. In our patient, the central corneal thickness was 600 micron in right eye and 580 micron in left eye. Therefore, when measuring and assessing the intraocular pressure, the influence of corneal thickness should be taken into consideration. One study reported that microspherophakia with loose zonules is more likely to dislocate than in cases of Marfan syndrome and happens in a downward direction¹⁹. Another study stated that lens dislocation in these patients occurs as frequently as in patients with Marfan syndrome and

homocystinuria, and glaucoma is more common than in either of the latter two conditions¹¹. Similar to other researchers, our patient was managed using the same strategy.

Conclusion:

Most cases of Weill Marchesani syndrome remain undiagnosed, often being treated as myopic eyes until complications such as secondary glaucoma and lens dislocation occur. Awareness and early diagnosis is important to prevent complications and thus blindness. Proper patient education, genetic counselling and screening of family members are also an essential part of management of this condition. In our case report, early diagnosis and timely proper management helped to regain functional vision with control of intraocular pressure.

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